



Missouri Department of Health and Senior Services

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Gail Vasterling
Director



Jeremiah W. (Jay) Nixon
Governor

August 3, 2015

LSD Newborn Screening in Missouri Goes Live

Dear Practitioner, Laboratory Manager, Newborn Nursery Manager:

In response to Missouri Statute RSMo 191.333.1, known as The Brady Alan Cunningham Newborn Screening Act, the Missouri Department of Health and Senior Services' Newborn Screening Program launched a full population pilot implementation process to screen for five Lysosomal Storage Disorders (LSDs) on all Missouri newborns. Krabbe Disease piloting was started in August of 2012, and then Pompe Disease, Gaucher Disease, Fabry Disease and Hurler Syndrome piloting commenced in January of 2013. The full population pilot phase for LSDs has been very successful.

Effective August 3rd, 2015, the Missouri State Public Health Laboratory (MSPHL) will be going live with the routine reporting of LSDs, thereby adding this as an additional disorder category to the standard newborn screening laboratory reports. High risk abnormal screening results will be phoned and faxed to the healthcare provider of record and to our contracted genetic referral centers who will follow up regarding confirmatory testing and next steps.

LSDs are genetic disorders resulting in enzyme deficiencies within the lysosomes of the body's cells. Lysosomes are the microscopic recycle bins in the cells that store, break down and recycle large unwanted molecules by utilizing very specific enzymes. The presence of an LSD results in an accumulation of these large molecules in the lysosomes resulting in their dysfunction, and ultimately irreversible damage to muscles, nerves and/or certain organs in the body. There are treatments available for these disorders if caught early before symptoms begin to appear in the infant.

Please note that providing the baby's gestational age and birth weight on the newborn screening collection form is vital to the MSPHL regarding screening result interpretations for LSDs.

If you have questions or comments, please contact: Patrick Hopkins, Chief of Newborn Screening Laboratory, 573-751-2662, or by email at Patrick.Hopkins@health.mo.gov

www.health.mo.gov

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